DiGeorge Syndrome/
Velocardiofacial Syndrome

Symptoms or Behaviors

- Wide-set eyes
- Low-set ears, with a notched ear fold
- Small jaw
- A narrow, short groove in the upper lip
- Cleft palate
- Recurrent infections, such as chronic runny nose or multiple bouts of pneumonia, oral thrush (candidiasis), diaper rash or diarrhea
- Cramp-like spasms of the baby's hands and fingers, or twitches or spasms of the muscles in the baby's face, throat or arms (tetany)
- Developmental delays, most often speech delay
- Slow mental development
- Lack of appetite
- Poor weight gain
- Failure to thrive

About the Disorder

What is DiGeorge syndrome?
DiGeorge syndrome is a disorder present at birth (congenital) caused by problems within a developing embryo that occur due to chromosome defects. It may also be identified as velocardiofacial syndrome.

Possible complications associated with DiGeorge syndrome are related to the specific medical problems caused by the chromosome defect.

If the parathyroid glands aren't fully developed (hypoparathyroidism), the child may experience stunted growth (short stature), slow mental development (or mental retardation), or cataracts.

If the thymus gland is underdeveloped or absent, severe infection may occur due to a lack of infection-fighting T cells.

Heart defects can result in severe complications including: congestive heart failure, enlargement of the heart (cardiomegaly), high blood pressure in the lungs (pulmonary hypertension), endocarditis or stroke.

How is DiGeorge Syndrome treated?
There is no cure for DiGeorge syndrome, and the long-term outlook for children with DiGeorge syndrome varies widely, depending on the organs affected. Treatment of DiGeorge syndrome focuses on correcting or managing the medical problems caused by the chromosome defect.
Educational Considerations

The information outlined below is related to Velo-Cardio-Facial Syndrome.

Developmental delay is a common manifestation in children with VCFS, but it is variable with some children showing almost no early problems and others being quite delayed. The delay in motor milestones has many contributors, including the generalized hypotonia (low muscle tone) that is common in VCFS. Also contributing are factors such as congenital heart disease, multiple hospitalizations and operations, chronic illness, and other factors that can restrict a child’s activity. By school age, the majority of children with VCFS tend to perform close to the normal range in terms of motor skills, although they may always be a little more hypotonic or even "clutzier" than other kids. Speech and language milestones tend to be slightly more delayed, with the average age of onset of the first word at 19 months, but again this is very variable. It would seem that children with VCFS have their own developmental profile that differs from that of other children. In general, most show significant "catch-up" before their fourth birthday.

To date, essentially all individuals with VCFS have shown some type of learning disorder. Although mental retardation can occur in VCFS, the diagnosis of mental retardation is reserved for a very small percentage of cases. Difficulty with abstraction and problem solving tends to interfere most with mathematics and reading comprehension. Concepts are difficult to grasp, even things as seemingly basic as "opposite" or "reverse." Therefore, learning is best done by repetition and breaking things down into the smallest concrete units.

Resources

Mayo Clinic
http://www.mayoclinic.com/health/digeorge-syndrome/DS00998

March of Dimes Birth Defects Foundation
1275 Mamaroneck Avenue, White Plains, NY 10605
Tel: (914)428-7100, Fax: (914)997-4763,
Tel: (888)663-4637
Email: Askus@marchofdimes.com
Internet: http://www.marchofdimes.com

Velo-Cardio-Facial Syndrome
Educational Foundation, Inc.
P.O. Box 874, Milltown, NJ 08850
Toll Free: 1-866-VCFSEF5 (1-866-823-7335)

Immune Deficiency Foundation
40 West Chesapeake Avenue, Suite 308
Towson, MD 21230
Tel: (410)321-6647, Fax: (410)321-9165
Tel: (800)296-4433
Email: idf@primaryimmune.org
Internet: http://www.primaryimmune.org

NIH/National Institute on Deafness and Other Communication Disorders (Balance)
National Temporal Bone, Hearing and Balance Pathology Resource Registry
Massachusetts Eye & Ear Infirmary
243 Charles Street, Boston, MA 02114-3096
Fax: (617)573-3838, Tel: (800)822-1327
TDD: (888)561-3277
Email: TBRegistry@meei.harvard.edu
Internet: http://www.tbregistry.org

NIH/National Institute of Child Health and Human Development
31 Center Dr, Building 31, Room 2A32, MSC2425
Bethesda, MD 20892
Tel: (301)496-5133
Fax: (301)496-7101
Internet: http://www.nih.gov/hichd/